

"V(D)J recombinase signal sequences in Alu elements" M. Richard Shen, Deborah A. Wilcox and Harvey W. Mohrenweiser, Biology and Biotechnology Research Program, L-452, LLNL, Livermore, CA 94550.

Alu elements contain a sequence that is very similar to the consensus V(D)J recombinase signal sequence (RSS). The Alu RSS matches the consensus RSS better in some Alu subfamilies than in others. We show that the Y (CS) Alu subfamily contains a RSS that may act as a RSS with either a 12 bp or a 23 bp spacer. The relative activity of the Y subfamily RSS as a 12 bp spacer is 0.5 ± 0.7 % of the consensus RSS, and is 0.2 ± 0.1 % as a 23 bp spacer. There are approximately 40,000 Y Alu elements per haploid human genome. However, there is a equal number of individual Alu elements that match the consensus RSS more closely than the Y Alu subfamily. Alu elements are frequently found in introns and 3' untranslated regions of genes. In the lymphocytic leukemias, these Alu RSSs, provide the potential for the formation of fusion proteins and exchanges of promoter elements between genes. There also remains the potential that a few Alu elements may have been exapted, to act as RSS, for programmed rearrangements within the primate genome. Research supported by LDRD 96-LW-091 to M.R.S. Work performed under auspices of the US DOE by the Lawrence Livermore National Laboratory; contract No. W-7405-ENG-48